

## Article

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## Abraham Lincoln, aortic regurgitation and abscesses

*by Douglas Hammond, Sunil Bhatia, Manoj Brahmabhatt, Almoi'zzlideenillah Mustafa and Jennifer Perry*

Abraham Lincoln was an exceptionally tall man with a long, thin face and huge hands and feet. His mother had a similar build – an observation that in 1962 led AM Gordon to first propose that President Lincoln had Marfan syndrome.<sup>1</sup>

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In 1972 Harold Schwartz published an article that described a seven-year-old patient with Marfan syndrome who was distantly related to Abraham Lincoln. The child had ancestry that was traced back to Mordecai Lincoln II, who was the great-great grandfather of Abraham Lincoln.<sup>2</sup> Lincoln also made the observation that he had blurry feet in photos, and the journalist who had taken the photographs suggested that the motion may be caused by the throbbing of the arteries in his leg. This gave rise to the hypothesis that President Lincoln had severe aortic regurgitation, which is found in Marfan syndrome.<sup>3</sup> This may be indicative of Traube's sign.

In his later years, Lincoln was also noted to have nodded his head on every heartbeat. In photos, owing to the increased shutter time on cameras of that age, his face appeared blurred compared with those around him. This is indicative of de Musset's sign, a condition named unusually after a patient rather than a doctor. It was named after the French poet Alfred de Musset, who died from syphilitic aortic regurgitation. Both of these signs are pathognomonic of aortic regurgitation, which is part of Marfan syndrome.

Without the ability to test the remains of Abraham Lincoln for DNA, it is impossible to tell whether Lincoln had the mutation within his genes. An alternative hypothesis is that he had Multiple Endocrine Neoplasia type 2B (MEN2B). This is a syndrome that is characterised by medullary thyroid cancer, pheochromocytomas, mucosal neuromas and a marfanoid habitus.<sup>4</sup> A long, thin face and prominent chin are thought to be typical marfanoid features.<sup>4</sup> Also both Abraham and his mother Nancy had melancholic expressions that are indicative of muscular hypotonia, a feature of MEN2B. Nancy passed away at the age of 34, which suggests that she died from a malignant process. MEN2B has multiple neoplastic elements, supporting the argument for this theory rather than Marfan syndrome.

#### Case report

A 25-year-old man attended the accident and emergency department with a rapidly enlarging sebaceous cyst on his forehead. He had this incised and drained under local anaesthesia by the DF2 trainee, and he was given a five-day course of oral antibiotics and an outpatient review appointment some months later.

In the outpatient department, he reported intermittent swelling of the cyst in the intervening months and requested excision. A full medical history was taken. When asked about any previous hospital admissions, he reported that he had had four previous pneumothoraces.

The attending clinician thought this unusual and this raised suspicion.

The patient was 195cm tall and, as the previous multiple pneumothoraces had aroused suspicion, he was examined and found to have a high arched palate, malocclusion, hypermobility of his thumbs, hyperelasticity of the skin and a resting tremor of his leg. When examined further, he was found to have a diastolic murmur consistent with aortic regurgitation and the distance between his outstretched arms was 1.1 times his height.

The patient was referred urgently to his GP for further investigation. When he returned to clinic for the uneventful excision of his sebaceous cyst under local anaesthesia, he reported that he had had ultrasonography of the aorta and echocardiography of the heart, which showed aortic root enlargement and aortic regurgitation. He had been referred urgently by his GP to the cardiology team. The patient had never met his father. However, his mother confirmed that his father had Marfan syndrome but that she had never sought medical screening for her son.

#### Discussion

Marfan syndrome is an autosomal dominant connective tissue disease described initially by Antoine Marfan in 1896.<sup>5</sup> It is classically described as being caused by mutations of the fibrillin-1 gene leading to abnormal synthesis, secretion or matrix incorporation of fibrillin, which is a glycoprotein essential for the formation of elastic fibres in connective tissue. However, more recent research suggests that many aspects of the disease are caused by altered regulation of transforming growth factor beta (TGF- $\beta$ ).<sup>6</sup> This highlights the potential of medical management of the syndrome by the therapeutic usage of TGF- $\beta$  antagonists.

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The prevalence of Marfan syndrome is about 2–3 per 10,000 individuals and affects males and females equally. This estimate depends on complete recognition of all affected and genetically predisposed individuals and, for the following reasons, its incidence may be underestimated:

- The older the patient the more prominent the phenotype, and therefore the more recognisable the condition becomes.
- The outward manifestations of the condition are common. Therefore clinicians may miss the diagnostic significance of these manifestations.
- Marfan syndrome cases can occur as *de novo* mutations. In 25% of cases this is true. Therefore not all patients present with a positive family history.
- There is no diagnostic test with a high sensitivity and specificity.<sup>7</sup>

Marfan syndrome is a multisystem condition and affects:

- **Skeleton:** Marfan syndrome patients are affected within their long bones. Hence they are tall and slender. Because of this the long bones, hands, fingers and toes are out of proportion with the rest of the body. They are also loose jointed. Intraorally, these individuals often have obvious signs, which are a high arched palate and dental crowding.
- **Cardiovascular system:** The atrioventricular valves are most often affected. The enlargement and thickening of atrioventricular valves is common. This is

often associated with prolapse of atrioventricular valves, and in some cases the tricuspid valves also.

- **Ocular:** More than 50% of people with Marfan syndrome have dislocation of one or both lenses of the eye.
- **Skin:** Patients with Marfan syndrome typically have normal skin texture but it is hyperelastic. The most common manifestations in the skin are *striae atrophicae* and hernias.<sup>8</sup>
- **Pulmonary system:** There are several reasons that patients with Marfan syndrome can have pulmonary disease. A restrictive pattern of lung disease can be caused by *Pectus excavatum* or progressive scoliosis.<sup>9</sup> Pneumothoraces can be caused by widening of the distal airspaces and formation of discrete bullae. These can easily lead to spontaneous pneumothorax. This is found in around 15% of patients with Marfan syndrome.<sup>10</sup>

In the 1970s patients with Marfan Syndrome had a reduced lifespan. It was thought that the lifespan of a patient with Marfan syndrome was about half to two-thirds of that of a person without Marfan syndrome.<sup>11</sup> This was mainly thought to be due to the risk of Aortic Dissection. In the 1970s the outcome for any patient with an aortic dissection was poor, and because Marfan syndrome predisposes people to this condition, life expectancy is frequently poor. However, there have been improvements in both the detection of Marfan syndrome and its complications, and also the treatment of aortic dissection. This has led to a more recent study showing that patients with Marfan syndrome have an almost-normal life expectancy.<sup>12</sup>

#### Learning points

It is well documented that the general dental practitioner has a major role in health screening for oral cancer<sup>13</sup> but risk assessment in general dental practice of medical conditions is also becoming increasingly common.<sup>14</sup> This has led to the development of care protocols, which aim to act as a framework for decision making to produce an optimum level of care. In practice, a significant proportion of patients attend on a non-symptomatic, continuous and regular basis, often over long periods of time. This provides general dental practitioners with a wealth of knowledge about their patients with which they can make informed clinical decisions on an individual basis.

As Sir William Osler famously stated, 'Listen to your patient, he is telling you the diagnosis.' A thorough history can lead to prompt detection of idiopathic conditions that may benefit from early diagnosis. In this case, the history allied with an examination of the fully clothed patient led to suspicion.



This case demonstrates that as a dental practitioner in either primary or secondary care, you are in a position to facilitate appropriate referral to either primary or secondary medical care. An ability to examine and listen to the heart is not entirely necessary to make the diagnosis. The suspicion arising from the multiple pneumothoraces as well as the presence of a high arched palate, malocclusion, hypermobility of the thumbs, hyperelasticity of the skin and a resting tremor of the leg, should be enough to prompt a referral.

Patients and dentists are both receptive to the idea of health screening at the dentist. Creanor *et al* found that 87% of patients thought that it was important or very important that dentists screened patients for medical conditions such as diabetes and 79% were very willing to let a dental team member carry out screening.<sup>15</sup> The majority indicated willingness to be screened for various medical conditions during a visit to the dentist, including hypertension and diabetes.

Dentists' attitudes are also changing. Greenberg *et al* found that the majority of dentists thought it was important to conduct screening for hypertension (85.8%), cardiovascular disease (76.8%), diabetes mellitus (76.6%), hepatitis (71.5%) and human immunodeficiency virus infection (68.8%).<sup>16</sup> Furthermore, 96.4% of dentists in the same study were also willing to refer patients for consultation with a medical practitioner.

As patients and dentists both feel that health screening in the clinic would be valuable, it may be time to incorporate this into practice. However, additional education and practical implementation strategies are necessary to bring this to fruition.

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